Robert W Taylor

List of Publications by Year
in descending order

351 papers	27,261 citations	7251 80 h-index	9865 146 g-index
356 all docs	356 docs citations	356 times ranked	24369 citing authors

#	Article	IF	Citations
1	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	3.7	25
2	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	1.4	12
3	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	2.8	17
4	The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. Journal of Raman Spectroscopy, 2022, 53, 172-181.	1.2	5
5	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062.	1.4	3
6	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
7	Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Analyst, The, 2022, 147, 2533-2540.	1.7	9
8	Defining mitochondrial protein functions through deep multiomic profiling. Nature, 2022, 606, 382-388.	13.7	49
9	Neuromuscular Junction Abnormalities in Mitochondrial Disease. Neurology: Clinical Practice, 2021, 11, 97-104.	0.8	10
10	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	5.8	21
11	The molecular pathology of pathogenic mitochondrial tRNA variants. FEBS Letters, 2021, 595, 1003-1024.	1.3	29
12	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	2.1	33
13	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	1.0	1
14	<scp>SLC25A42</scp>â€associated mitochondrial encephalomyopathy: Report of additional founder cases and functional characterization of a novel deletion. JIMD Reports, 2021, 60, 75-87.	0.7	6
15	Machine learning algorithms reveal the secrets of mitochondrial dynamics. EMBO Molecular Medicine, 2021, 13, e14316.	3.3	6
16	A novel MT-CO2 variant causing cerebellar ataxia and neuropathy: The role of muscle biopsy in diagnosis and defining pathogenicity. Neuromuscular Disorders, 2021, 31, 1186-1193.	0.3	5
17	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	4.9	96
18	Interrogating Mitochondrial Biology and Disease Using CRISPR/Cas9 Gene Editing. Genes, 2021, 12, 1604.	1.0	10

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19	The Effect of tRNA[Ser]Sec Isopentenylation on Selenoprotein Expression. International Journal of Molecular Sciences, 2021, 22, 11454.	1.8	8
20	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	3.3	5
21	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	1.7	113
22	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
23	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	1.1	14
24	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286.	1.0	7
25	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	2.6	39
26	Assessment of mitochondrial respiratory chain enzymes in cells and tissues. Methods in Cell Biology, 2020, 155, 121-156.	0.5	32
27	Lewy body pathology is more prevalent in older individuals with mitochondrial disease than controls. Acta Neuropathologica, 2020, 139, 219-221.	3.9	11
28	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. Neuromuscular Disorders, 2020, 30, 661-668.	0.3	8
29	Earlyâ€onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic COQ8A variants, including a large intragenic deletion. JIMD Reports, 2020, 54, 45-53.	0.7	8
30	SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. Molecular Genetics and Metabolism Reports, 2020, 25, 100657.	0.4	10
31	<scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659.	3.3	44
32	Nuclear genetic disorders of mitochondrial DNA gene expression. , 2020, , 375-409.		0
33	The genetic basis of isolated mitochondrial complex II deficiency. Molecular Genetics and Metabolism, 2020, 131, 53-65.	0.5	22
34	Age-associated mitochondrial DNA mutations cause metabolic remodeling that contributes to accelerated intestinal tumorigenesis. Nature Cancer, 2020, 1, 976-989.	5.7	69
35	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	3.8	48
36	Mitochondrial OXPHOS Biogenesis: Co-Regulation of Protein Synthesis, Import, and Assembly Pathways. International Journal of Molecular Sciences, 2020, 21, 3820.	1.8	74

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37	Novel MT-ND Gene Variants Causing Adult-Onset Mitochondrial Disease and Isolated Complex I Deficiency. Frontiers in Genetics, 2020, 11, 24.	1.1	14
38	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	3.3	45
39	Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial tRNA ^{Gly} (MT-TG) variant. Neurology: Genetics, 2020, 6, e413.	0.9	2
40	Progressive external ophthalmoplegia due to a recurrent de novo m.15990C>T MT-TP (mt-tRNAPro) gene variant. Neuromuscular Disorders, 2020, 30, 346-350.	0.3	4
41	Chronic Progressive External Ophthalmoplegia due to a Rare de novo m.12334G>A MT-TL2 Mitochondrial DNA Variant1. Journal of Neuromuscular Diseases, 2020, 7, 355-360.	1.1	2
42	Biâ€allelic pathogenic variants in NDUFC2 cause earlyâ€onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619.	3.3	17
43	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	1.4	19
44	A novel mitochondrial m.4414T>C MT-TM gene variant causing progressive external ophthalmoplegia and myopathy. Neuromuscular Disorders, 2019, 29, 693-697.	0.3	2
45	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	1.4	19
46	Resolving complexity in mitochondrial disease: Towards precision medicine. Molecular Genetics and Metabolism, 2019, 128, 19-29.	0.5	25
47	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. Cell Reports, 2019, 26, 996-1009.e4.	2.9	116
48	A Novel Pathogenic Variant in MT-CO2 Causes an Isolated Mitochondrial Complex IV Deficiency and Late-Onset Cerebellar Ataxia. Journal of Clinical Medicine, 2019, 8, 789.	1.0	11
49	Cognitive deficits in adult m.3243A>Gâ€and m.8344A>Gâ€related mitochondrial disease: importance of correcting for baseline intellectual ability. Annals of Clinical and Translational Neurology, 2019, 6, 826-836.	1.7	10
50	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443.	6.5	16
51	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
52	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
53	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2057-2066.	1.8	19
54	A novel pathogenic m.4412G>A MT-TM mitochondrial DNA variant associated with childhood-onset seizures, myopathy and bilateral basal ganglia changes. Mitochondrion, 2019, 47, 18-23.	1.6	4

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55	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	1.7	17
56	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108.	1.6	12
57	Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. Nature Communications, 2019, 10, 759.	5.8	34
58	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
59	Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219.	1.3	26
60	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	1.1	31
61	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	2.6	59
62	Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127.	0.7	36
63	Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. Scientific Reports, 2018, 8, 1799.	1.6	30
64	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	1.1	50
65	Disclosing the functional changes of two genetic alterations in a patient with Chronic Progressive External Ophthalmoplegia: Report of the novel mtDNA m.7486G>A variant. Neuromuscular Disorders, 2018, 28, 350-360.	0.3	10
66	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	2.8	42
67	Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6.	4.5	102
68	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
69	Loss-of-function mutations inISCA2disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. Human Mutation, 2018, 39, 537-549.	1.1	21
70	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345.	1.7	102
71	Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73.	0.5	25
72	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	1.5	73

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73	NEW OBSERVATIONS REGARDING THE RETINOPATHY OF GENETICALLY CONFIRMED KEARNSâ€“SAYRE SYNDROME. Retinal Cases and Brief Reports, 2018, 12, 349-358.	0.3	8
74	A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 Câ€methyltransferase deficiency. Human Mutation, 2018, 39, 69-79.	1.1	43
75	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
76	Expanding the clinical phenotype of IARS2-related mitochondrial disease. BMC Medical Genetics, 2018, 19, 196.	2.1	16
77	A Wars2 Mutant Mouse Model Displays OXPHOS Deficiencies and Activation of Tissue-Specific Stress Response Pathways. Cell Reports, 2018, 25, 3315-3328.e6.	2.9	35
78	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	2.6	41
79	Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. Human Molecular Genetics, 2018, 27, 4135-4144.	1.4	30
80	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	3.3	54
81	Expanding the phenotype of de novo SLC25A4 -linked mitochondrial disease to include mild myopathy. Neurology: Genetics, 2018, 4, e256.	0.9	20
82	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. Annals of Neurology, 2018, 84, 289-301.	2.8	47
83	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	3.3	199
84	Confirming TDP2 mutation in spinocerebellar ataxia autosomal recessive 23 (SCAR23). Neurology: Genetics, 2018, 4, e262.	0.9	27
85	Inherited pathogenic mitochondrial DNA mutations and gastrointestinal stem cell populations. Journal of Pathology, 2018, 246, 427-432.	2.1	13
86	POLG2 deficiency causes adultâ€onset syndromic sensory neuropathy, ataxia and parkinsonism. Annals of Clinical and Translational Neurology, 2017, 4, 4-14.	1.7	13
87	MR-1S Interacts with PET100 and PET117 in Module-Based Assembly of Human Cytochrome c Oxidase. Cell Reports, 2017, 18, 1727-1738.	2.9	86
88	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41
89	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	2.5	217
90	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	5.8	432

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91	Diabetes Mellitus in Mitochondrial Disease. Frontiers in Diabetes, 2017, , 55-68.	0.4	1
92	Clinically proven mtDNA mutations are not common in those with chronic fatigue syndrome. BMC Medical Genetics, 2017, 18, 29.	2.1	15
93	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	1.5	54
94	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
95	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	0.9	11
96	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	0.7	10
97	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	2.6	58
98	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	1.4	18
99	Pigmentary retinopathy, rodâ€“cone dysfunction and sensorineural deafness associated with a rare mitochondrial tRNALys(m.8340G>A) gene variant. British Journal of Ophthalmology, 2017, 101, 1298-1302.	2.1	8
100	Multipotent Basal Stem Cells, Maintained in Localized Proximal Niches, Support Directed Long-Ranging Epithelial Flows in Human Prostates. Cell Reports, 2017, 20, 1609-1622.	2.9	64
101	Novel POLG variants associated with late-onset de novo status epilepticus and progressive ataxia. Neurology: Genetics, 2017, 3, e181.	0.9	2
102	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. Scientific Reports, 2017, 7, 15676.	1.6	20
103	The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250.	2.1	329
104	Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130.	1.7	23
105	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	0.9	1
106	Nucleotide pools dictate the identity and frequency of ribonucleotide incorporation in mitochondrial DNA. PLoS Genetics, 2017, 13, e1006628.	1.5	55
107	Compound heterozygous RMND1 gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. BMC Research Notes, 2016, 9, 325.	0.6	15
108	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	2.6	57

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109	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113.	0.9	12
110	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	1.5	35
111	Incidence of Primary Mitochondrial Disease in Children Younger Than 2 Years Presenting With Acute Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2016, 63, 592-597.	0.9	40
112	Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131.	1.5	91
113	Clinical features of the pathogenic m.5540G>A mitochondrial transfer RNA tryptophan gene mutation. Neuromuscular Disorders, 2016, 26, 702-705.	0.3	6
114	Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
115	Impaired mitochondrial biogenesis is a common feature to myocardial hypertrophy and end-stage ischemic heart failure. Cardiovascular Pathology, 2016, 25, 103-112.	0.7	77
116	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. Nucleic Acids Research, 2016, 44, 5313-5329.	6.5	37
117	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	4.5	69
118	Dysferlin mutations and mitochondrial dysfunction. Neuromuscular Disorders, 2016, 26, 782-788.	0.3	28
119	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	2.6	93
120	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	4.5	241
121	Mitochondrial dysfunction in myofibrillar myopathy. Neuromuscular Disorders, 2016, 26, 691-701.	0.3	32
122	Investigating complex <scp>I</scp> deficiency in <scp>P</scp>urkinje cells and synapses in patients with mitochondrial disease. Neuropathology and Applied Neurobiology, 2016, 42, 477-492.	1.8	23
123	Lethal Neonatal LTBL Associated with Biallelic EARS2 Variants: Case Report and Review of the Reported Neuroradiological Features. JIMD Reports, 2016, 33, 61-68.	0.7	23
124	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	1.6	165
125	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	2.8	40
126	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. Nature Communications, 2016, 7, 12317.	5.8	106

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127	The swinging pendulum of biomarkers in mitochondrial disease. Neurology, 2016, 87, 2286-2287.	1.5	5
128	A recurrent mitochondrial p.Trp22ArgNDUFB3variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. Journal of Medical Genetics, 2016, 53, 634-641.	1.5	31
129	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	2.6	89
130	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	3.7	15
131	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	1.7	92
132	Short peptides from leucyl-tRNA synthetase rescue disease-causing mitochondrial tRNA point mutations. Human Molecular Genetics, 2016, 25, 903-915.	1.4	19
133	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
134	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. European Heart Journal, 2016, 37, 2552-2559.	1.0	53
135	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	1.7	79
136	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779.	1.5	67
137	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	2.8	62
138	A novel immunofluorescent assay to investigate oxidative phosphorylation deficiency in mitochondrial myopathy: understanding mechanisms and improving diagnosis. Scientific Reports, 2015, 5, 15037.	1.6	104
139	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
140	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 151-155.	1.1	8
141	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
142	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a C12orf65 Related Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 409-419.	1.1	22
143	Triplex real-time PCRâ€“an improved method to detect a wide spectrum of mitochondrial DNA deletions in single cells. Scientific Reports, 2015, 5, 9906.	1.6	30
144	A recessive homozygous p.Asp92Gly SDHD mutation causes prenatal cardiomyopathy and a severe mitochondrial complex II deficiency. Human Genetics, 2015, 134, 869-879.	1.8	49

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145	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
146	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	4.5	41
147	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
148	Neuropathologic Characterization of Pontocerebellar Hypoplasia Type 6 Associated With Cardiomyopathy and Hydrops Fetalis and Severe Multisystem Respiratory Chain Deficiency due to NovelRARS2Mutations. Journal of Neuropathology and Experimental Neurology, 2015, 74, 688-703.	0.9	31
149	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular Disorders, 2015, 25, 262-267.	0.3	9
150	Mitochondrial Donation â€" How Many Women Could Benefit?. New England Journal of Medicine, 2015, 372, 885-887.	13.9	87
151	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. Frontiers in Genetics, 2015, 6, 21.	1.1	46
152	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. Clinical Science, 2015, 128, 895-904.	1.8	21
153	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2015, 97, 319-328.	2.6	83
154	A novel m.7539C>T point mutation in the mt-tRNAAsp gene associated with multisystemic mitochondrial disease. Neuromuscular Disorders, 2015, 25, 81-84.	0.3	10
155	Long-term survival in a child with severe encephalopathy, multiple respiratory chain deficiency and GFM1 mutations. Frontiers in Genetics, 2015, 6, 102.	1.1	13
156	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	2.6	110
157	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	7.2	119
158	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	1.7	45
159	Mutations causing mitochondrial disease: What is new and what challenges remain?. Science, 2015, 349, 1494-1499.	6.0	251
160	The presence of highly disruptive 16S rRNA mutations in clinical samples indicates a wider role for mutations of the mitochondrial ribosome in human disease. Mitochondrion, 2015, 25, 17-27.	1.6	29
161	Mitochondrial and inflammatory changes in sporadic inclusion body myositis. Neuropathology and Applied Neurobiology, 2015, 41, 288-303.	1.8	73
162	Periventricular Calcification, Abnormal Pterins and Dry Thickened Skin: Expanding the Clinical Spectrum of RMND1?. JIMD Reports, 2015, 26, 13-19.	0.7	6

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163	Pathogenic mitochondrial mt-tRNAAla variants are uniquely associated with isolated myopathy. European Journal of Human Genetics, 2015, 23, 1735-1738.	1.4	24
164	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. European Journal of Human Genetics, 2015, 23, 935-939.	1.4	32
165	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	2.6	41
166	SANDO syndrome in a cohort of 107 patients with CPEO and mitochondrial DNA deletions. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 630-634.	0.9	113
167	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	1.1	19
168	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
169	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. European Journal of Human Genetics, 2014, 22, 184-191.	1.4	52
170	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	1.1	64
171	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	1.4	57
172	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. PLoS Genetics, 2014, 10, e1004424.	1.5	112
173	Clonal Expansion of Early to Mid-Life Mitochondrial DNA Point Mutations Drives Mitochondrial Dysfunction during Human Ageing. PLoS Genetics, 2014, 10, e1004620.	1.5	115
174	Bmi1 enhances skeletal muscle regeneration through MT1-mediated oxidative stress protection in a mouse model of dystrophinopathy. Journal of Experimental Medicine, 2014, 211, 2617-2633.	4.2	34
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